



ETHE1 gene

ETHE1, persulfide dioxygenase

Normal Function

The *ETHE1* gene provides instructions for making an enzyme that is involved in energy production. This enzyme is active throughout the body in mitochondria (the energy-producing centers in cells), where it may form part of a larger protein complex. Little is known about the enzyme's function in mitochondria.

Health Conditions Related to Genetic Changes

ethylmalonic encephalopathy

More than 20 different mutations in the *ETHE1* gene have been identified in people with ethylmalonic encephalopathy. Most of these mutations lead to the production of an abnormally short, nonfunctional version of the ETHE1 enzyme or prevent cells from making any of this enzyme. These genetic changes impair the body's ability to produce energy in mitochondria. Additionally, a lack of the ETHE1 enzyme allows potentially toxic compounds, including ethylmalonic acid and lactic acid, to build up in the body. Excess amounts of these compounds can be detected in urine.

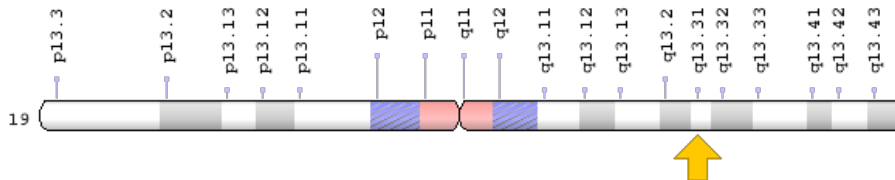
It is unclear how a loss of the ETHE1 enzyme leads to the progressive features of ethylmalonic encephalopathy. Researchers suggest that a buildup of toxic compounds in the brain could be responsible for the neurologic signs and symptoms. Changes in the blood vessels or in blood flow could also contribute to nervous system damage, leading to the characteristic features of the disorder.

Leigh syndrome

Chromosomal Location

Cytogenetic Location: 19q13.31, which is the long (q) arm of chromosome 19 at position 13.31

Molecular Location: base pairs 43,506,719 to 43,527,256 on chromosome 19 (Homo sapiens Annotation Release 108, GRCh38.p7) (NCBI)



Credit: Genome Decoration Page/NCBI

Other Names for This Gene

- ETHE1_HUMAN
- ethylmalonic encephalopathy 1
- Ethylmalonic encephalopathy protein 1
- hepatoma subtracted clone one
- HSCO
- YF13H12

Additional Information & Resources

Scientific Articles on PubMed

- PubMed
<https://www.ncbi.nlm.nih.gov/pubmed?term=%28%28ETHE1%5BTIAB%5D%29%29+OR+%28ethylmalonic+encephalopathy+1%5BTIAB%5D%29%29+OR+%28hepatoma+subtracted+clone+one%5BTIAB%5D%29%29+OR+%28HSCO%5BTIAB%5D%29%29+OR+%28YF13H12%5BTIAB%5D%29%29+OR+%28Ethylmalonic+encephalopathy+protein+1%5BTIAB%5D%29%29+AND+%28%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+3600+days%22%5Bdp%5D>

OMIM

- ETHE1 GENE
<http://omim.org/entry/608451>

Research Resources

- ClinVar
<https://www.ncbi.nlm.nih.gov/clinvar?term=ETHE1%5Bgene%5D>
- HGNC Gene Symbol Report
http://www.genenames.org/cgi-bin/gene_symbol_report?q=data/hgnc_data.php&hgnc_id=23287
- NCBI Gene
<https://www.ncbi.nlm.nih.gov/gene/23474>
- UniProt
<http://www.uniprot.org/uniprot/O95571>

Sources for This Summary

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Reviewed: August 2006

Published: March 21, 2017

Lister Hill National Center for Biomedical Communications
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